

Variant: NM_005633.3(SOS1):c.2988G>A (p.Pro996=)

CA136123 [↗](#)

40713 (ClinVar) [↗](#)

Gene: SOS1 ([HGNC:6654](#))

Condition: RASopathy ([MONDO:0021060](#))

Inheritance Mode: Autosomal dominant inheritance

UID: 47edbaff-50ac-42ee-838b-2ac0c4a4054c

Approved on: 2017-04-18

Published on: 2018-12-10

HGVS expressions

NM_005633.3:c.2988G>A

NM_005633.3(SOS1):c.2988G>A (p.Pro996=)

NC_000002.12:g.38997015C>T

CM000664.2:g.38997015C>T

NC_000002.11:g.39224156C>T

CM000664.1:g.39224156C>T

NC_000002.10:g.39077660C>T

NG_007530.1:g.128449G>A

ENST00000395038.6:c.2988G>A

ENST00000402219.6:c.2988G>A

ENST00000426016.5:c.2988G>A

Benign

Met criteria codes **1**

BA1

Evidence Links **0**

Expert Panel

[RASopathy VCEP](#) [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

RASopathy VCEP

The filtering allele frequency of the c.2988G>A (p.Pro996=) variant in the SOS1 gene is 0.396% (51/10072) of African chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BA1; PMID:29493581)

Met criteria codes

BA1



The filtering allele frequency of the c.2988G>A (p.Pro996=) variant in the SOS1 gene is 0.396% (51/10072) of African chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BA1; PMID:29493581)

Curation History [↗](#)



Showing 1 to 1 of 1 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.